

HUMAN CHROMOSOME 15 AND 16 BARDET-BIEDL SYNDROME
POLYNUCLEOTIDES AND POLYPEPTIDES AND METHODS OF USE

ABSTRACT OF THE DISCLOSURE

Amino acid and nucleic acid sequences of Bardet-Biedl Syndrome Region (BBSR) that map to human chromosome 15 or 16 are disclosed. Compositions and methods related to the genes and proteins are useful in the study, diagnosis and treatment of a variety of diseases including BBS and related conditions including obesity, retinal degeneration, and disorders affecting the nervous system, the heart, the kidneys, and the like. Compositions include those comprising BBSR polypeptides and derivatives thereof, nucleotide sequences, expression cassettes, vectors, transformed cells and antibodies. Methods include those for expression and detection of BBSR nucleotides and polypeptides.